deafness and myopia syndrome

Deafness and myopia syndrome is a disorder that causes problems with both hearing and vision. People with this disorder have moderate to profound hearing loss in both ears that may worsen over time. The hearing loss may be described as sensorineural, meaning that it is related to changes in the inner ear, or it may be caused by auditory neuropathy, which is a problem with the transmission of sound (auditory) signals from the inner ear to the brain. The hearing loss is either present at birth (congenital) or begins in infancy, before the child learns to speak (prelingual).

Affected individuals also have severe nearsightedness (high myopia). These individuals are able to see nearby objects clearly, but objects that are farther away appear blurry. The myopia is usually diagnosed by early childhood.

Frequency

The prevalence of deafness and myopia syndrome is unknown. Only a few affected families have been described in the medical literature.

Genetic Changes

Deafness and myopia syndrome is caused by mutations in the *SLITRK6* gene. The protein produced from this gene is found primarily in the inner ear and the eye. This protein promotes growth and survival of nerve cells (neurons) in the inner ear that transmit auditory signals. It also controls (regulates) the growth of the eye after birth. In particular, the SLITRK6 protein influences the length of the eyeball (axial length), which affects whether a person will be nearsighted or farsighted, or will have normal vision. The SLITRK6 protein spans the cell membrane, where it is anchored in the proper position to perform its function.

SLITRK6 gene mutations that cause deafness and myopia syndrome result in an abnormally short SLITRK6 protein that is not anchored properly to the cell membrane. As a result, the protein is unable to function normally. Impaired SLITRK6 protein function leads to abnormal nerve development in the inner ear and improperly controlled eyeball growth, resulting in the hearing loss and nearsightedness that occur in deafness and myopia syndrome.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- deafness and myopia
- deafness, cochlear, plus
- DFNMYP
- high myopia and sensorineural deafness
- high myopia-sensorineural deafness syndrome
- myopia and deafness

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Deafness and myopia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1857342/

Other Diagnosis and Management Resources

- Baby's First Test: Hearing Loss
  http://www.babysfirsttest.org/newborn-screening/conditions/hearing-loss
- EyeSmart: Eyeglasses for Vision Correction
  https://www.aao.org/eye-health/glasses-contacts/glasses
- GeneReview: Deafness and Myopia Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK269029
- Harvard Medical School Center for Hereditary Deafness
  http://hearing.harvard.edu/
- KidsHealth: Hearing Evaluation in Children
  http://kidshealth.org/en/parents/hear.html
- MedlinePlus Encyclopedia: Cochlear Implant
  https://medlineplus.gov/ency/article/007203.htm
- MedlinePlus Health Topic: Cochlear Implants
  https://medlineplus.gov/cochlearimplants.html
- MedlinePlus Health Topic: Hearing Aids
  https://medlineplus.gov/hearingaids.html
- MedlinePlus Health Topic: Newborn Screening
  https://medlineplus.gov/newbornscreening.html

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Cochlear Implant
  https://medlineplus.gov/ency/article/007203.htm
- Health Topic: Cochlear Implants
  https://medlineplus.gov/cochlearimplants.html
- Health Topic: Hearing Aids
  https://medlineplus.gov/hearingaids.html
- Health Topic: Hearing Problems in Children
  https://medlineplus.gov/hearingproblemsinchildren.html
- Health Topic: Newborn Screening
  https://medlineplus.gov/newbornscreening.html
- Health Topic: Refractive Errors
  https://medlineplus.gov/refractiveerrors.html

Genetic and Rare Diseases Information Center

- Deafness and myopia syndrome
  https://rarediseases.info.nih.gov/diseases/12844/deafness-and-myopia-syndrome

Additional NIH Resources

- National Eye Institute (NEI): Myopia
  https://nei.nih.gov/health/errors/myopia
- National Institute on Deafness and Other Communication Disorders (NIDCD): Auditory Neuropathy
  https://www.nidcd.nih.gov/health/auditory-neuropathy

Educational Resources

- American Academy of Otolaryngology--Head and Neck Surgery: Genes and Hearing Loss
  http://www.entnet.org/content/genes-and-hearing-loss
- Center for Hearing and Speech: Types of Hearing Loss
  http://www.centerhearingandspeech.org/types-hearing-loss/
- Centers for Disease Control and Prevention: Hearing Loss in Children -- Types of Hearing Loss
  https://www.cdc.gov/NCBDDD/hearingloss/types.html
- KidsHealth: Auditory Neuropathy Spectrum Disorder
  http://kidshealth.org/en/parents/ansd.html
- KidsHealth: Your Child's Vision
  http://kidshealth.org/en/parents/vision.html
- Laurent Clerc National Deaf Education Center
  http://www3.gallaudet.edu/clerc-center.html
- MalaCards: deafness and myopia
  http://www.malacards.org/card/deafness_and_myopia
- My Baby's Hearing: Auditory Neuropathy
  https://www.babyhearing.org/hearingamplification/causes/neuropathy.asp
- Orphanet: High myopia-sensorineural deafness syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=363396
- University of Arizona: Myopia and Deafness
  http://disorders.eyes.arizona.edu/disorders/myopia-and-deafness

Patient Support and Advocacy Resources

- Alexander Graham Bell Center for the Deaf and Hard of Hearing
  https://www.agbell.org/Default.aspx
- March of Dimes: Hearing Loss
  http://www.marchofdimes.org/baby/hearing-impairment.aspx
- National Association of the Deaf
  https://www.nad.org/
- Royal National Institute of Blind People: Myopia and High Degree Myopia
  http://www.rnib.org.uk/eye-health-eye-conditions-z-eye-conditions/myopia-and-high-degree-myopia
- University of Kansas Genetics Education Center Resource List: Hard of Hearing/ Deafness
  http://www.kumc.edu/gec/support/hearing.html

GeneReviews

- Deafness and Myopia Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK269029

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28deafness+and+myopia+syndrome%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- DEAFNESS AND MYOPIA
  http://omim.org/entry/221200

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